



## EDA gene

ectodysplasin A

### Normal Function

The *EDA* gene provides instructions for making a protein called ectodysplasin A. This protein is part of a signaling pathway that plays an important role in development before birth. Specifically, it is critical for interactions between two embryonic cell layers called the ectoderm and the mesoderm. In the early embryo, these cell layers form the basis for many of the body's organs and tissues. Ectoderm-mesoderm interactions are essential for the formation of several structures that arise from the ectoderm, including the skin, hair, nails, teeth, and sweat glands.

The *EDA* gene provides instructions for producing many slightly different versions of ectodysplasin A. One version, ectodysplasin A1, interacts with a protein called the ectodysplasin A receptor (produced from the *EDAR* gene). On the cell surface, ectodysplasin A1 attaches to this receptor like a key in a lock. When these two proteins are connected, they trigger a series of chemical signals that affect cell activities such as division, growth, and maturation. Before birth, this signaling pathway controls the formation of ectodermal structures such as hair follicles, sweat glands, and teeth.

### Health Conditions Related to Genetic Changes

#### hypohidrotic ectodermal dysplasia

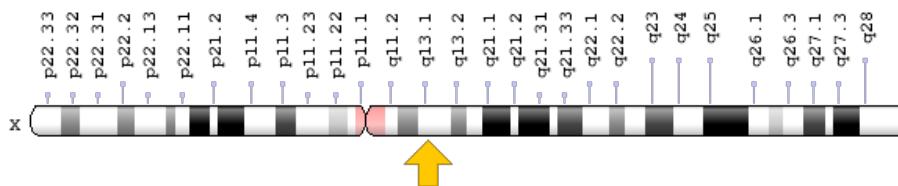
More than 80 different mutations in the *EDA* gene have been identified in people with hypohidrotic ectodermal dysplasia. These mutations cause the X-linked form of the disorder, which accounts for 95 percent of all cases of hypohidrotic ectodermal dysplasia. (X-linked disorders are caused by mutations in genes on the X chromosome, one of the two sex chromosomes.)

Some mutations in the *EDA* gene change single DNA building blocks (base pairs), whereas other mutations insert or delete genetic material in the gene. These changes lead to the production of a nonfunctional version of the ectodysplasin A protein. This abnormal protein cannot trigger chemical signals needed for normal interactions between the ectoderm and the mesoderm. Without these signals, hair follicles, teeth, sweat glands, and other ectodermal structures do not form properly, leading to the characteristic features of hypohidrotic ectodermal dysplasia.

## Chromosomal Location

Cytogenetic Location: Xq13.1, which is the long (q) arm of the X chromosome at position 13.1

Molecular Location: base pairs 69,616,067 to 70,039,472 on the X chromosome (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- Ectodermal dysplasia protein
- ectodysplasin
- ectodysplasin-A
- ED1
- ED1-A1
- EDA-A1
- EDA-A2
- EDA1
- EDA\_HUMAN
- HED
- XHED
- XLHED

## Additional Information & Resources

### Educational Resources

- Eda/Edar Signaling (Eurekah Bioscience Collection)  
<https://www.ncbi.nlm.nih.gov/books/NBK6103/>

## GeneReviews

- Hypohidrotic Ectodermal Dysplasia  
<https://www.ncbi.nlm.nih.gov/books/NBK1112>

## Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28EDA%5BTIAB%5D%29+OR%28ectodysplasin+A%5BTIAB%5D%29%29+OR+%28%28ectodysplasin-A%5BTIAB%5D%29+OR+%28ED1%5BTIAB%5D%29+OR+%28EDA1%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D>

## OMIM

- ECTODYSPASIN A  
<http://omim.org/entry/300451>

## Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_EDA.html](http://atlasgeneticsoncology.org/Genes/GC_EDA.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=EDA%5Bgene%5D>
- HGNC Gene Family: Tumor necrosis factor superfamily  
<http://www.genenames.org/cgi-bin/genefamilies/set/781>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=3157](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=3157)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/1896>
- UniProt  
<http://www.uniprot.org/uniprot/Q92838>

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<https://ghr.nlm.nih.gov/gene/EDA>

Reviewed: August 2006

Published: March 21, 2017

Lister Hill National Center for Biomedical Communications  
U.S. National Library of Medicine  
National Institutes of Health  
Department of Health & Human Services